

# **Methylmalonic Acidemia (MMA)**

An organic acid disorder

## ***What is it?***

Methylmalonic Acidemia (also known as MMA) is an inherited organic acid disorder. People with organic acid disorders, like MMA, cannot properly break down certain components of protein and fats. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the proteins and fats, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

## ***What are the symptoms?***

A person with MMA can appear normal at birth. The symptoms of MMA can be very variable between people. Some people with MMA will have the following symptoms after a few days of life: poor feeding, lack of energy, vomiting, low muscle tone, seizures, and trouble breathing. Kidney failure may develop. People with MMA may also present with the following symptoms later in infancy: failure to thrive, developmental delay, and seizures. People with MMA may have no symptoms at all. Many symptoms of MMA can be prevented by immediate treatment and lifelong management. People with MMA typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

## ***Inheritance and frequency***

MMA is inherited in an autosomal recessive manner. This means that for a person to be affected with MMA, he or she must have inherited two non-working copies of the gene responsible for causing MMA. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have MMA. Typically, there is no family history of MMA in an affected person. About 1 in 50,000 babies born have MMA.

## ***How is it detected?***

MMA can be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

## ***How is it treated?***

MMA is treated by eating a diet low in protein and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

**DISCLAIMER:** This information is not intended to replace the advice of a genetic metabolic medical professional.

**For more information:**

**Genetics Home Reference**

Website: <http://ghr.nlm.nih.gov/ghr/page/Home>

**Save Babies Through Screening Foundation**

4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545 Email: [email@savebabies.org](mailto:email@savebabies.org)

Website: <http://www.savebabies.org/diseasedescriptions.php/>

**Organic Acidemia Association**

13210 - 35th Avenue North Plymouth, MN 55441 **Phone:** 763-559-

1797 **Fax:** 763-694-0017 **Email:** [oaanews@aol.com](mailto:oaanews@aol.com)

[www.oaanews.org](http://www.oaanews.org)

**STAR-G Hawaii Department of Health**

<http://www.newbornscreening.info/Parents/organicaciddisorders/MMA.html>